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What is claimed is:

1. Isolated nucleic acid encoding a human EHOC-1 polypeptide.
2. Isolated nucleic acid according to claim 1, wherein said nucleic acid comprises DNA.
3. DNA according to claim 2, wherein said DNA is a cDNA.
4. DNA according to claim 2, wherein said DNA encodes the amino acid sequence set forth in SEQ ID NO: 2.
5. DNA according to claim 2, wherein said DNA hybridizes under high stringency conditions to substantially the entire coding sequence (nucleotides 157-3726) set forth in SEQ ID NO: 2.
6. DNA according to claim 2, wherein said DNA has substantially the same nucleotide sequence as the nucleotide sequence set forth in SEQ ID NO: 1.
7. A vector comprising DNA according to claim 2.
8. A host cell containing a vector according to claim 7, wherein said cell is a procaryotic cell or a eucaryotic cell.
9. A host cell according to claim 8, wherein said cell expresses a functional EHOC-1 protein.
10. A nucleic acid probe comprising at least 15 nucleotides capable of specifically hybridizing with a sequence of nucleic acids of the nucleotide sequence set forth in SEQ ID NO: 1.

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12. A kit for detecting mutations and aneuploidies in chromosome 21 at locus q22.3 comprising a plurality of probes, wherein each probe comprises a nucleic acid sequence having at least 15 bp of contiguous nucleotides capable of specifically hybridizing with a sequence of nucleic acids of the nucleotide sequence set forth in SEQ ID NO: 1, and wherein each individual probe corresponds to a specific locus on chromosome 21q22.3

14. An oligonucleotide composition comprising chemical analogues of the nucleic acid of claim 2 operatively linked to a promoter of RNA transcription.

16. Isolated EHOC-1 polypeptide and functional equivalents thereof.

18. Isolated EHOC-1 polypeptide according to claim 16, wherein said polypeptide has the same amino acid sequence as that set forth in SEQ ID NO: 2.

19. Isolated EHOC-1 polypeptide according to claim 16, wherein said polypeptide is encoded by a

nucleotide sequence that is substantially the same nucleotide sequence as that set forth in SEQ ID NO: 1.

20. Isolated EHOC-1 polypeptide according to claim 16, wherein said polypeptide is encoded by the nucleotide sequence set forth in SEQ ID NO: 1.

21. An EHOC-1 polypeptide expressed recombinantly in a host cell.

22. An EHOC-1 polypeptide according to claim 21, wherein said polypeptide is encoded by a nucleotide sequence that is substantially the same as the nucleotide sequence set forth in SEQ ID NO: 1.

23. An EHOC-1 polypeptide according to claim 21, wherein said polypeptide is encoded by the nucleotide sequence set forth in SEQ ID NO: 1.

24. An antibody that specifically binds to a determinant on a human EHOC-1 protein or active fragment thereof.

25. An antibody according to claim 24, wherein said antibody is a monoclonal antibody.

26. An antibody according to claim 24, wherein said antibody is a polyclonal antibody.

27. A composition comprising an amount of the antisense oligonucleotide according to claim 13 effective to modulate expression of a human EHOC-1 polypeptide and an acceptable hydrophobic carrier capable of passing through a cell membrane.

28. A composition according to claim 27, wherein the oligonucleotide is coupled to a substance which inactivates mRNA.

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37. A method for identifying nucleic acids encoding a human EHOC-1 protein, said method comprising:

contacting a sample containing nucleic acids with a probe according to claim 11, wherein said contacting is effected under high stringency hybridization conditions, and identifying compounds which hybridize thereto.

38. A method for identifying compound(s) which bind to a human EHOC-1 polypeptide, said method comprising contacting cells according to claim 9 with said compound(s) and identifying compounds which bind thereto.

39. A method for detecting the presence of a human EHOC-1 polypeptide on a cell surface, said method comprising contacting a test cell with an antibody according to claim 24, detecting the presence of an antibody-receptor complex, and therefor detecting the presence of a human EHOC-1 polypeptide on the cell surface.

40. A method for diagnosing a predisposition to a disorder associated with the expression of a specific human EHOC-1 polypeptide allele, said method comprising:

contacting a sample containing nucleic acids with a plurality of probes, wherein each probe comprises a nucleic acid sequence having at least 15 bp of contiguous nucleotides capable of specifically hybridizing with a sequence of nucleic acids of the nucleotide sequence set forth in SEQ ID NO: 1, and wherein each individual probe corresponds to a specific locus on chromosome 21q22.3

41. A method according to claim 40, wherein said disorder is selected from progressive myoclonus epilepsy, holoprosencephaly, or autoimmune polyglandular disease.

42. A method for deterring the onset of symptoms associated with particular disorder comprising

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administering a composition which modulates expression of gene.

43. A method for introducing changes at human chromosome locus 21q22.3 comprising transforming a sample of cells obtained from a subject having progressive myoclonus epilepsy with the nucleic acid according to claim 1 along with a selective marker gene; maintaining cells in selective media; and

isolating viable cells containing a modified target sequence.

44. A method of supplying wild-type EHOC-1 gene function to a cell which has a mutation/aneuploidy in the EHOC-1 gene comprising introducing a wild-type EHOC-1 gene or functional fragment thereof into said cell such that it is expressed.

45. Single strand DNA primers for amplification diagnosis of progressive myoclonus epilepsy, wherein said primers comprise a nucleic acid sequence derived from the nucleic acid sequence set forth as SEQ ID NO: 1.

46. A method for detecting one or more EHOC-1 alleles in a sample of nucleic acid comprising determining the presence or absence of variant nucleotide sequence in a gene contained in any of BAC clones.

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